

Disease

Congenital sucrase-isomaltase deficiency^{1, 2, 3}

Gene

SI

Inheritance	AR
Current Treatments	Avoid sucrose and isomaltose. Oral sacrosidase
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	13C-sucrose labeled breath test ⁴
Disease Frequency (per 100,000)	110 ^{5, 6}
Genes Included in Disease Frequency	SI
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=sucrase&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/222900>

² <https://pubmed.ncbi.nlm.nih.gov/33772704/>

³ <https://pubmed.ncbi.nlm.nih.gov/33567694/>

⁴ <https://pubmed.ncbi.nlm.nih.gov/19330928/>

⁵ <https://bmcpediatr.biomedcentral.com/articles/10.1186/1471-2431-14-11>

⁶ <https://ghr.nlm.nih.gov/condition/congenital-sucrase-isomaltase-deficiency#synonyms>